Journal of Forensic and Legal Medicine 20 (2013) 520–521

Contents lists available at SciVerse ScienceDirect

Journal of Forensic and Legal Medicine

journal homepage: www.elsevier.com/locate/jflm

Short report

Delta-storage pool disease as a mimic of abusive head trauma in a 7-month-old baby: A case report

Marc De Leeuw MD [a], Emile Beuls MD [a], Philippe Jorens MD, PhD [b], Paul Parizel MD, PhD [c], Werner Jacobs MD, PhD, Prof., Head of Department of Forensic Medicine and Pathology [a,*]

[a] Department of Forensic Medicine and Pathology, University Hospital Antwerp, Wilrijkstraat 10, B-2650 Edegem, Belgium
[b] Department of Intensive Care Medicine, University Hospital Antwerp, Edegem, Belgium
[c] Department of Radiology, University Hospital Antwerp, Edegem, Belgium

ARTICLE INFO

Article history:
Received 6 August 2012
Received in revised form
30 January 2013
Accepted 3 March 2013
Available online 11 April 2013

Keywords:
Shaken baby syndrome
Abusive head trauma
Mimic
Delta-storage pool disease

ABSTRACT

A seven-month-old baby was admitted to a hospital emergency department after collapsing suddenly while staying with his nanny. The baby displayed classic symptoms of shaken baby syndrome, including subdural haemorrhage, cytotoxic cerebral oedema, and bilateral retinal hemorrhages. Child protection services were informed, but both the parents and the nanny denied any involvement. In the subsequent weeks, the baby developed three other episodes of new subdural bleeding and a medico-legal investigation was started into the origin of the repeated subdural bleeding. Eventually, platelet aggregation tests and electron microscopy diagnosed a delta-storage pool disease; that is, a haemostatic disorder involving dense granules of the platelets. Initial minor blunt trauma may have resulted in subdural bleeding, while subsequent retinal haemorrhage could have been facilitated by the underlying haemostatic disorder. Delta-storage pool disease should be considered as a possible mimic of abusive head trauma similar to other rare conditions such as Menkes disease and type 1 glutaric aciduria.

1. Introduction

Child abusive head trauma continues to be one of the most significant challenges in clinical and forensic medicine. Clinical physicians must strike a fine balance between the child's safety and the risk of falsely accusing the child's caregivers. Forensic physicians have to advise judicial authorities as to whether there is sufficient scientific and forensic evidence to prosecute a person suspected of having committed a serious crime against a young child. There is an adage that it is better to release 100 guilty people than to falsely convict one innocent person. However, child abusers rarely confess their crimes. Starling et al.[1] identified babysitters as an important risk group for child abusive head trauma. Therefore, forensic physicians and pathologists often carry the burden of providing the scientific proof for the crime. The presence of classic clusters of clinical symptoms with 95% diagnostic certainty might not be enough for forensic or judicial purposes. In the absence of a confession, the medico-legal work-out of the case should be stringent and consider all alternative possibilities, including vary rare conditions, before a judicial authority can be advised, beyond reasonable doubt, that the case is truly one of abusive head trauma. In the past, most paediatric and forensic medicine handbooks and review papers mentioned three classic but rare medical conditions that had to be excluded before abusive head trauma was diagnosed in the absence of a confession: osteogenesis imperfecta, Menkes disease and type 1 glutaric aciduria.[2,3] In 1991, however, Tsementsis and Marsh[4] described for the first time the association between a platelet storage pool disease and spontaneous subarachnoid haemorrhage. The present case report illustrates the case history of a child suspected to be the victim of repetitive abusive head trauma, but with underlying platelet disorder. We argue that delta-storage pool disease, a platelet disorder, should be added to this list of potential medical pitfalls of abusive head trauma.

2. Case description

A 7-month-old male baby was trusted to the care of a nanny when, after lunch, he suddenly lost consciousness and became hypotonic. The baby was resuscitated by a medical response team and transferred to a university hospital, where he was diagnosed with an extensive subdural haematoma. The magnetic resonance imaging (MRI) scan demonstrated multifocal cytotoxic oedema.

* Corresponding author.
E-mail address: werner.jacobs@uza.be (W. Jacobs).

1752-928X/$ – see front matter © 2013 Elsevier Ltd and Faculty of Forensic and Legal Medicine. All rights reserved.
http://dx.doi.org/10.1016/j.jflm.2013.03.021

No external bruising or fractures were seen. Routine blood testing for bleeding and metabolic disorders was normal, but 5 days after admission at the intensive care ward, the baby developed a deep venous thrombosis of the left femoral vein. The child was born *a terme* after an uncomplicated pregnancy and there was no significant medical history in the family. Only frequent night-time crying was reported. Since the baby also displayed bilateral retinal haemorrhages, a case of shaken baby syndrome was suspected and child protection services were informed. Both parents and the nanny – the only people who had access to the child – were questioned by the police, but all denied any involvement. The child remained under the care of both parents.

Twelve weeks later, a control MRI scan led to a diagnosis of a new subdural bleeding in the old bleeding with compression of the brain parenchyma. At this time, the parents did not mention any specific symptoms. An emergent neurosurgical evacuation was performed.

Another 12 weeks later, the child was admitted to the hospital because of severe vomiting. Medical imaging again showed mixed-density subdural bleeding with punctiform frontal bleeding. Extensive testing for bleeding disorders (von Willebrand, haemophilia A/B/C, genetic predisposition for thrombosis, abnormal prothrombin variant, etc.) was started, which eventually turned out to be negative. Cerebral angiography to exclude vasculopathy such as Moya–Moya disease was considered. Genetic counselling was negative for hereditary bleeding disorders. Clinical stigmata for the autosomal recessive disorder Hermanski–Pudlak syndrome (HPS) were not present.

Almost 2 years after the initial incident, the diagnosis of delta-storage pool disease was made by electron microscopy. It was the medical opinion of the child haematologist that a delta-storage pool disease was not responsible for spontaneous intracranial bleeding, but that minor trauma could initiate a craniocerebral bleeding.

The forensic medical experts advised the court that this was not a typical case of child abusive head trauma and that there was insufficient evidence for a shaken baby syndrome. Although minor head trauma might have caused the primary event, all subsequent events could be explained by rebleeding due to the underlying disorder. A court ruling is still pending as of the time of writing. The child currently has a severe developmental retardation with spasticity in the limbs.

3. Discussion

Although neuroimaging is crucial for diagnosing child abuse and radiological findings can be highly suggestive, they often remain non-specific in the absence of other corroborating evidence or a confession. The source of potential pitfalls in the diagnosis of child abusive head trauma is a diagnostic minefield. Among the factors that should be considered are characteristics of skull fractures, normal prominent tentorium and falx versus subdural haematoma, birth trauma versus non-accidental head trauma, hyperacute versus acute chronic subdural haematomas and expanded subarachnoid space versus subdural haemorrhage. Further, controversies regarding subdural haematomas associated with benign enlarged subarachnoid spaces, hypoxia as a cause of subdural haematoma and/or retinal haemorrhages without trauma, the significance of retinal haemorrhages related to non-accidental head trauma, the significance of subdural haematomas in general and pitfalls of glutaric aciduria type 1 mimicking non-accidental head trauma[5] should be taken into account. Tung et al.[6] reported that homogeneous hyperdense subdural haematoma is more frequent in cases of accidental head trauma. Mixed-density subdural haematoma is more frequently seen in cases of non-accidental head injury, but may be observed within 48 h of accidental head trauma.

Delta-storage pool disease is a platelet storage pool deficiency related to deficient functioning of lysosomes and resulting in a bleeding tendency. Mutations in the HPS genes seem to be correlated with platelet dense-granule deficiency.[7] In delta-storage disease, delta granules (also named dense granules) are affected (few or new granules present). The platelet count is normal, but the bleeding time may be prolonged. A definite diagnosis is made using electron microscopy when few or no dense granules are demonstrated in the platelets. The absence of delta granules results in a deficient platelet plug formation and may cause a prolonged bleeding time. A study by Torres-Serrant et al.[8] found that 1.18% of newborn Puerto Rican children carried the mutated HPS-3 gene. Puerto Ricans seem to be a particularly at-risk population due to a 16-bp duplication in exon 15,[7] although other mutations have also been described in non-Puerto Ricans.

In a forensic setting, De Munnynck et al.[9] reported a case of repeated anal bleeding in a young child, which raised suspicions of repetitive anal sexual abuse. However, an underlying delta-storage pool disease was also present.

Rapid diagnosis of pool storage diseases might be desirable from both clinical and forensic perspectives. A diagnostic delay of almost 2 years would appear unacceptable in light of an ongoing police and judicial enquiry with a high risk that the child or other children will be removed from the family during that time for preventive protective reasons. However, the diagnosis is not made by routine or even special blood tests. The ultimate diagnostic tests are platelet aggregation tests and electron microscopy[10] demonstrating only few or no dense granules in the platelets. However, Maurer-Spurej et al.[11] reported a rapid 2-h flowcytometry test for platelet serotonin content in children, which was lower in delta-storage pool disease than in healthy children.

This case illustrates the importance of considering and diagnosing rare platelet disorders in cases of recurrent subdural bleeding suspected for repetitive abusive head trauma.

Ethical approval
None.

Funding
None.

Conflicts of interest
None.

References

1. Starling SP, Holden JR, Jenny C. Abusive head trauma: the relationship of perpetrators to their victims. *Pediatrics* 1995;**95**:259–62.
2. Menkes JH. Subdural haematoma, non-accidental head injury or …? *Eur J Paediatr Neurol* 2001;**5**:175–6.
3. Köhler M, Hoffmann GF. Subdural haematoma in a child with glutaric aciduria type I. *Pediatr Radiol* 1998;**28**:582.
4. Tsementzis SA, Marsh J. Spontaneous subarachnoid haemorrhage occurring in association with a platelet function disorder. *Neurosurg Rev* 1991;**14**:57–9.
5. Fernando S, Obaldo RE, Walsh IR, Lowe LH. Neuroimaging of nonaccidental head trauma: pitfalls and controversies. *Pediatr Radiol* 2008;**38**:827–38.
6. Tung GA, Kumar M, Richardson RC, Jenny C, Brown WD. Comparison of accidental and nonaccidental traumatic head injury in children on noncontrast computed tomography. *Pediatrics* 2006;**118**:626–33.
7. Huizing M, Gahl WA. Disorders of vesicles of lysosomal lineage: the Hermansky–Pudlak syndromes. *Curr Mol Med* 2002;**2**:451–67.
8. Torres-Serrant M, Ramirez SI, Cadilla CL, Ramos-Valencia G, Santiago-Borrero PJ. Newborn screening for Hermansky–Pudlak syndrome type 3 in Puerto Rico. *J Pediatr Hematol Oncol* 2012;**32**:448–53.
9. De Munnynck K, Van Geet C, De Vos R, Van De Voorde W. Delta-storage pool disease: a pitfall in the forensic investigation of sudden anal blood loss in children: a case report. *Int J Leg Med* 2007;**121**:44–7.
10. White JG. Use of the electron microscope for diagnosis of platelet disorders. *Semin Thromb Hemost* 1998;**24**:163–8.
11. Maurer-Spurej J, Pittendreigh C, Wu JK. Diagnosing platelet delta-storage pool disease in children by flow cytometry. *Am J Clin Pathol* 2007;**127**:626–32.